

ABSTRACT

The present invention relates to the identification of a gene, now designated negevin (*ngvn*), that is involved in the genetic disease Bardet Biedl Syndrome (BBS), which is characterized by such diverse symptoms as obesity, diabetes, hypertension, mental retardation, renal cancer and other abnormalities, retinopathy and hypogonadism. The human NGVN protein disclosed herein is 731 amino acids in length and is coded for by a gene spanning 17 exons. Homologs have been identified in mouse, rat, zebrafish. Methods of use for the gene, for example in diagnosis and therapy of BBS and in drug screening, also are described.